Turner syndrome

Turner syndrome is a chromosomal condition that affects development in females. The most common feature of Turner syndrome is short stature, which becomes evident by about age 5. An early loss of ovarian function (ovarian hypofunction or premature ovarian failure) is also very common. The ovaries develop normally at first, but egg cells (oocytes) usually die prematurely and most ovarian tissue degenerates before birth. Many affected girls do not undergo puberty unless they receive hormone therapy, and most are unable to conceive (infertile). A small percentage of females with Turner syndrome retain normal ovarian function through young adulthood.

About 30 percent of females with Turner syndrome have extra folds of skin on the neck (webbed neck), a low hairline at the back of the neck, puffiness or swelling (lymphedema) of the hands and feet, skeletal abnormalities, or kidney problems. One third to one half of individuals with Turner syndrome are born with a heart defect, such as a narrowing of the large artery leaving the heart (coarctation of the aorta) or abnormalities of the valve that connects the aorta with the heart (the aortic valve). Complications associated with these heart defects can be life-threatening.

Most girls and women with Turner syndrome have normal intelligence. Developmental delays, nonverbal learning disabilities, and behavioral problems are possible, although these characteristics vary among affected individuals.

Frequency

This condition occurs in about 1 in 2,500 newborn girls worldwide, but it is much more common among pregnancies that do not survive to term (miscarriages and stillbirths).

Genetic Changes

Turner syndrome is related to the X chromosome, which is one of the two sex chromosomes. People typically have two sex chromosomes in each cell: females have two X chromosomes, while males have one X chromosome and one Y chromosome. Turner syndrome results when one normal X chromosome is present in a female's cells and the other sex chromosome is missing or structurally altered. The missing genetic material affects development before and after birth.

About half of individuals with Turner syndrome have monosomy X, which means each cell in the individual's body has only one copy of the X chromosome instead of the usual two sex chromosomes. Turner syndrome can also occur if one of the sex chromosomes is partially missing or rearranged rather than completely absent. Some women with Turner syndrome have a chromosomal change in only some of their cells,

which is known as mosaicism. Women with Turner syndrome caused by X chromosome mosaicism are said to have mosaic Turner syndrome.

Researchers have not determined which genes on the X chromosome are associated with most of the features of Turner syndrome. They have, however, identified one gene called *SHOX* that is important for bone development and growth. The loss of one copy of this gene likely causes short stature and skeletal abnormalities in women with Turner syndrome.

Inheritance Pattern

Most cases of Turner syndrome are not inherited. When this condition results from monosomy X, the chromosomal abnormality occurs as a random event during the formation of reproductive cells (eggs and sperm) in the affected person's parent. An error in cell division called nondisjunction can result in reproductive cells with an abnormal number of chromosomes. For example, an egg or sperm cell may lose a sex chromosome as a result of nondisjunction. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have a single X chromosome in each cell and will be missing the other sex chromosome.

Mosaic Turner syndrome is also not inherited. In an affected individual, it occurs as a random event during cell division in early fetal development. As a result, some of an affected person's cells have the usual two sex chromosomes, and other cells have only one copy of the X chromosome. Other sex chromosome abnormalities are also possible in females with X chromosome mosaicism.

Rarely, Turner syndrome caused by a partial deletion of the X chromosome can be passed from one generation to the next.

Other Names for This Condition

- 45,X
- monosomy X
- TS
- Turner's syndrome
- Ullrich-Turner syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Turner syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0041408/

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Ovarian Hypofunction
  https://medlineplus.gov/ency/article/001163.htm
- MedlinePlus Encyclopedia: Turner Syndrome
  https://medlineplus.gov/ency/article/000379.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Ovarian Hypofunction
  https://medlineplus.gov/ency/article/001163.htm
- Encyclopedia: Turner Syndrome
  https://medlineplus.gov/ency/article/000379.htm
- Health Topic: Turner Syndrome
  https://medlineplus.gov/turnersyndrome.html

Genetic and Rare Diseases Information Center

- Turner syndrome
  https://rarediseases.info.nih.gov/diseases/7831/turner-syndrome

Additional NIH Resources

- Eunice Kennedy Shriver National Institute of Child Health and Human Development
  https://www.nichd.nih.gov/health/topics/turner/Pages/default.aspx
- National Human Genome Research Institute
  https://www.genome.gov/19519119/

Educational Resources

- Boston Children's Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/turner-syndrome
- Centre for Genetics Education (Australia)
  http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/FactSheet40TurnerSyndrome.pdf%20
- Disease InfoSearch: Turner syndrome
  http://www.diseaseinfosearch.org/Turner+syndrome/7268
- Genetic Science Learning Center, University of Utah
  http://learn.genetics.utah.edu/content/disorders/chromosomal/
- MalaCards: turner syndrome
  http://www.malacards.org/card/turner_syndrome
- March of Dimes: Chromosomal Conditions
  http://www.marchofdimes.org/baby/chromosomal-conditions.aspx
- Merck Manual Consumer Version
  http://www.merckmanuals.com/home/children-s-health-issues/chromosomal-and-genetic-abnormalities/turner-syndrome
- My46 Trait Profile
  https://www.my46.org/trait-document?trait=Turner%20syndrome&type=profile
- Orphanet: Turner syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=881
- TeensHealth from the Nemours Foundation
  http://kidshealth.org/en/teens/turner.html
- The MAGIC Foundation
  https://www.magicfoundation.org/Growth-Disorders/Turner-Syndrome/

Patient Support and Advocacy Resources

- National Organization for Rare Disorders
  https://rarediseases.org/rare-diseases/turner-syndrome/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/chromoso.html#xo
- Turner Syndrome Society of the United States
  http://www.turnersyndrome.org/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22turner+syndrome%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Turner+Syndrome%5BMAJR%5D%29+AND+%28Turner+syndrome%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

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